Medical Informatics Europe – MIE 2020,
HL7 International Virtual Workshop, 17 July 2020

Formal ontologies and multilingual terminologies as tools for knowledge level interoperability in the biomedical domain

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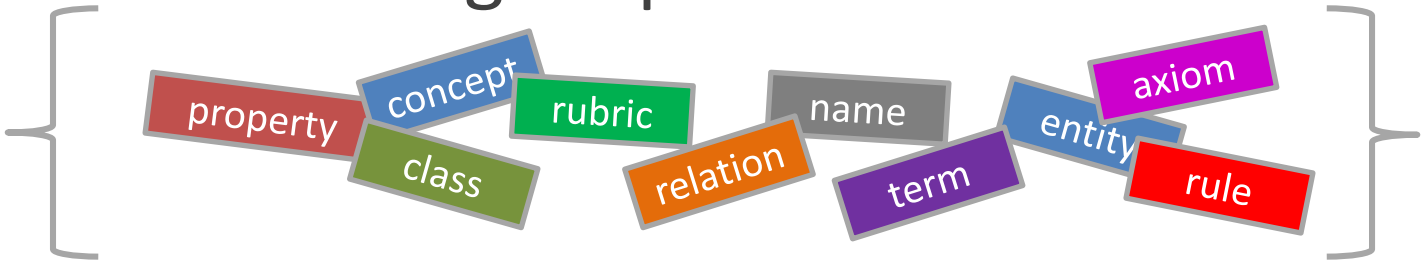
EFMI EUROPEAN FEDERATION for MEDICAL INFORMATICS

HL7 International

Problem

- Babylonian language confusion in biomedical semantics & knowledge representation

Knowledge



  - What kinds of knowledge need to be represented?
  - Is a more principled framework possible?
  - How do biomedical formal ontologies and multilingual terminologies fit in this picture?

Schulz S, Jansen L. Formal ontologies in biomedical knowledge representation. *Yearb Med Inform*. 2013;8:132-146.

Knowledge map

Universals

denotation

instantiation

"perro", "dog"
"canino", “Hund”
"canis", "dog"
NCBI:txid9615
"Marley"

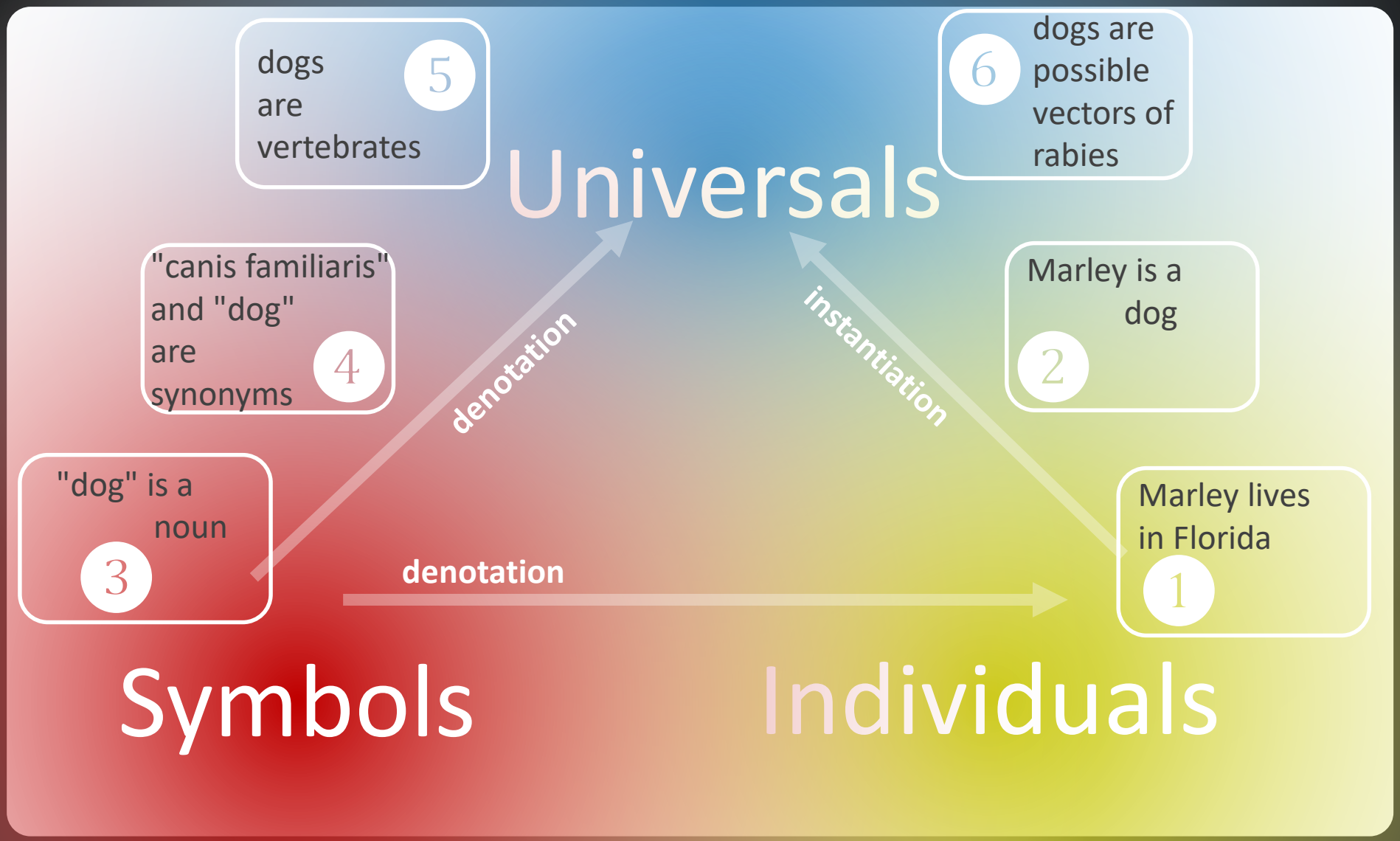
denotation

Symbols

Individuals

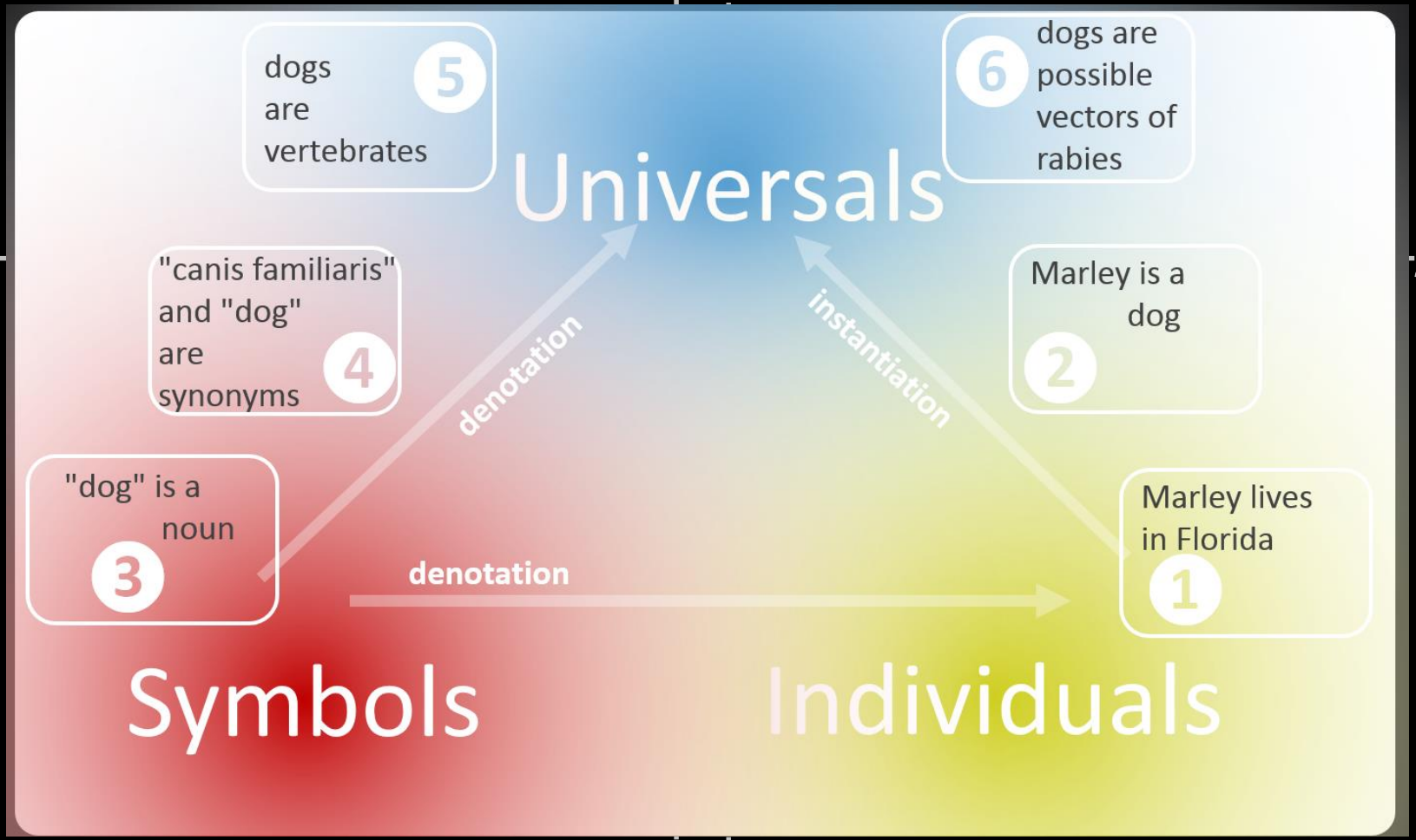
C. K. Ogden and I. A. Richards (1923) The Meaning of Meaning

Knowledge map



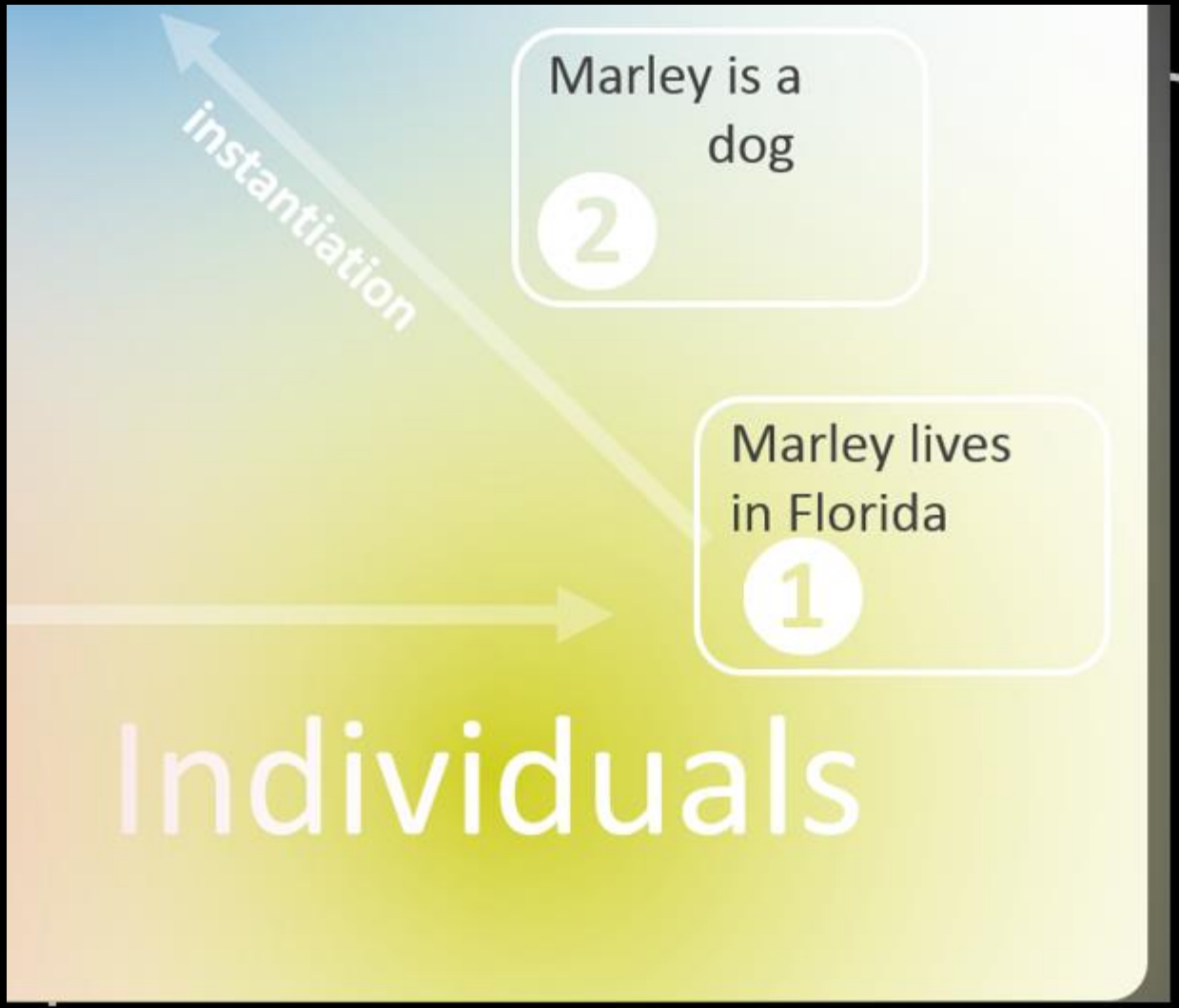
Ontological "knowledge":
Axioms that are universally true

Contingent knowledge:
typical, likely, possible



Symbolic knowledge:
Statements about properties and meaning of signs of language

Factual knowledge:
Statements about concrete entities and their relationships



Factual knowledge:
Statements about concrete entities and their relationships

Statements about individuals

<Subject> <Predicate> <Object>

```
:Florida rdf:type :state
:Marley rdf:type :dog
:Marley :lives :Florida
```

Syntax TURTLE : https://www.w3.org/TR/turtle/

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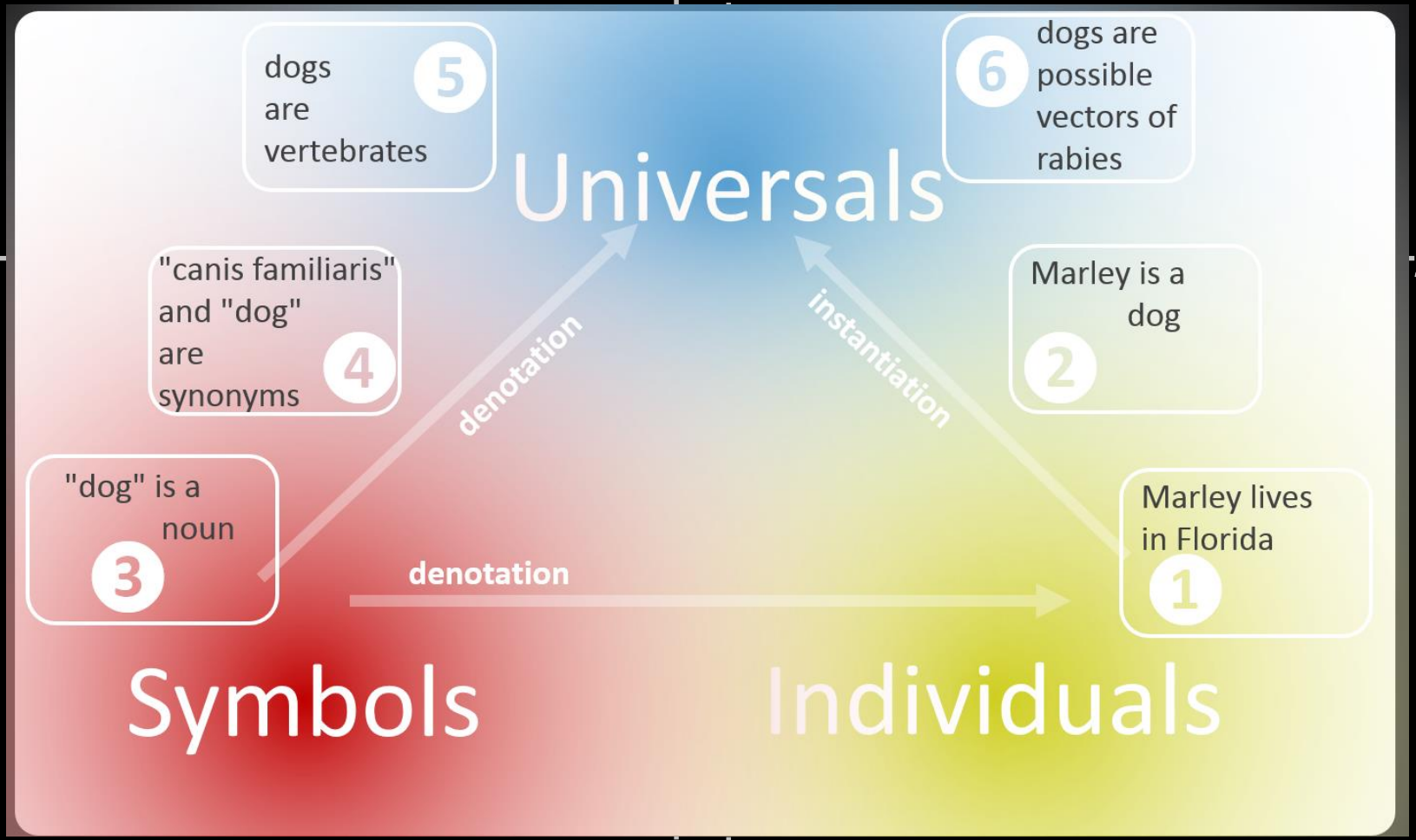

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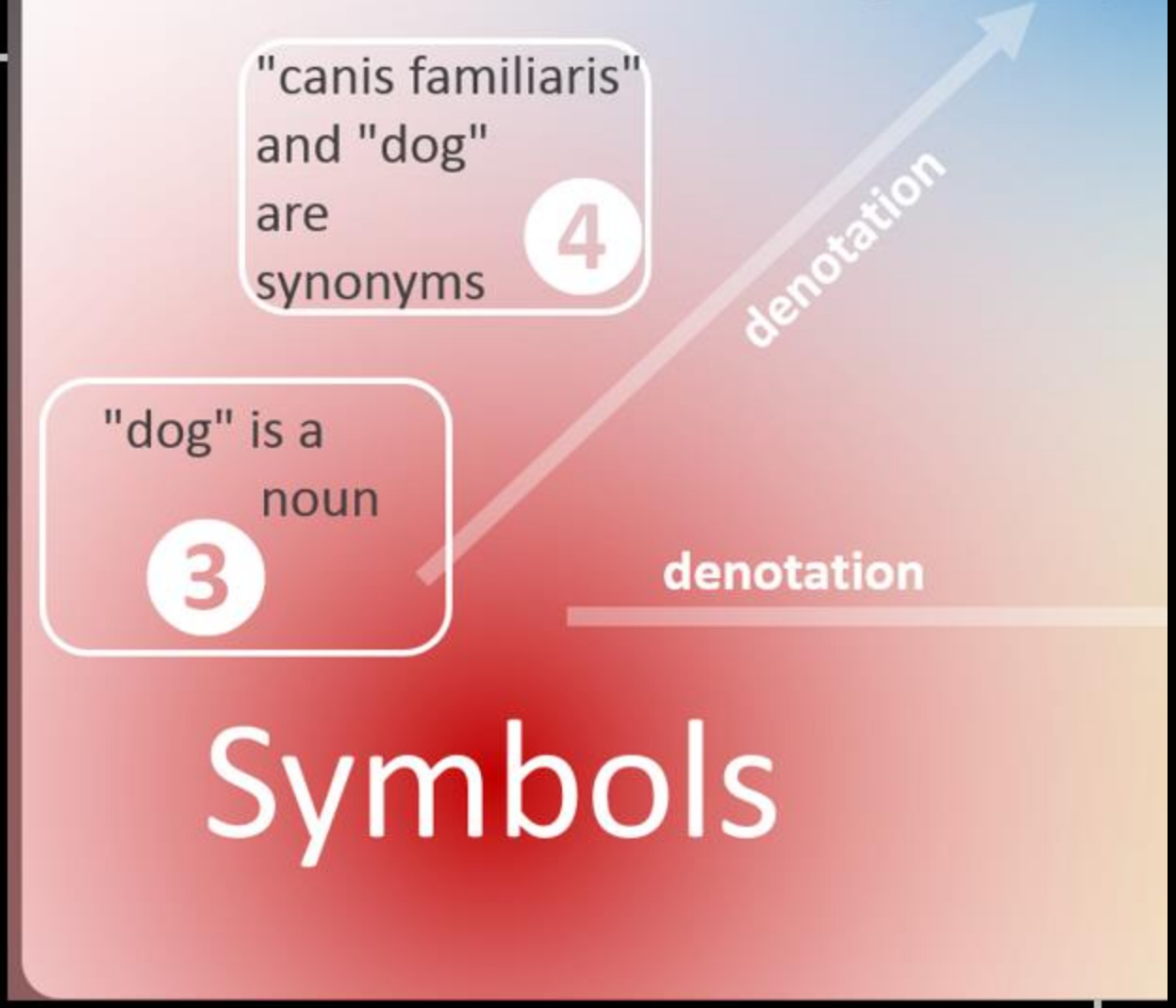

Symbolic knowledge

Statements about properties and meaning of signs of language

Representations SKOS / Linked Data

```
:ex:Dog rdf:type skos:Concept
:ex:Dog skos:prefLabel "dog"@en;
:ex:Dog skos:prefLabel "perro"@es;

:ex:Animal rdf:type skos:Concept
:ex:Animal skos:broader ex:Dog

wr:dog    lemon:sense    wr:dog-English-Noun-1
wr:dog    lemon:sense    wr:dog-English-Verb-1
wr:dog-English-Noun-1    wt:hasPoS wt:Noun
```

Syntax TURTLE : https://www.w3.org/TR/turtle/
Wiktionary: http://wiki.dbpedia.org/wiktionary-rdf-extraction

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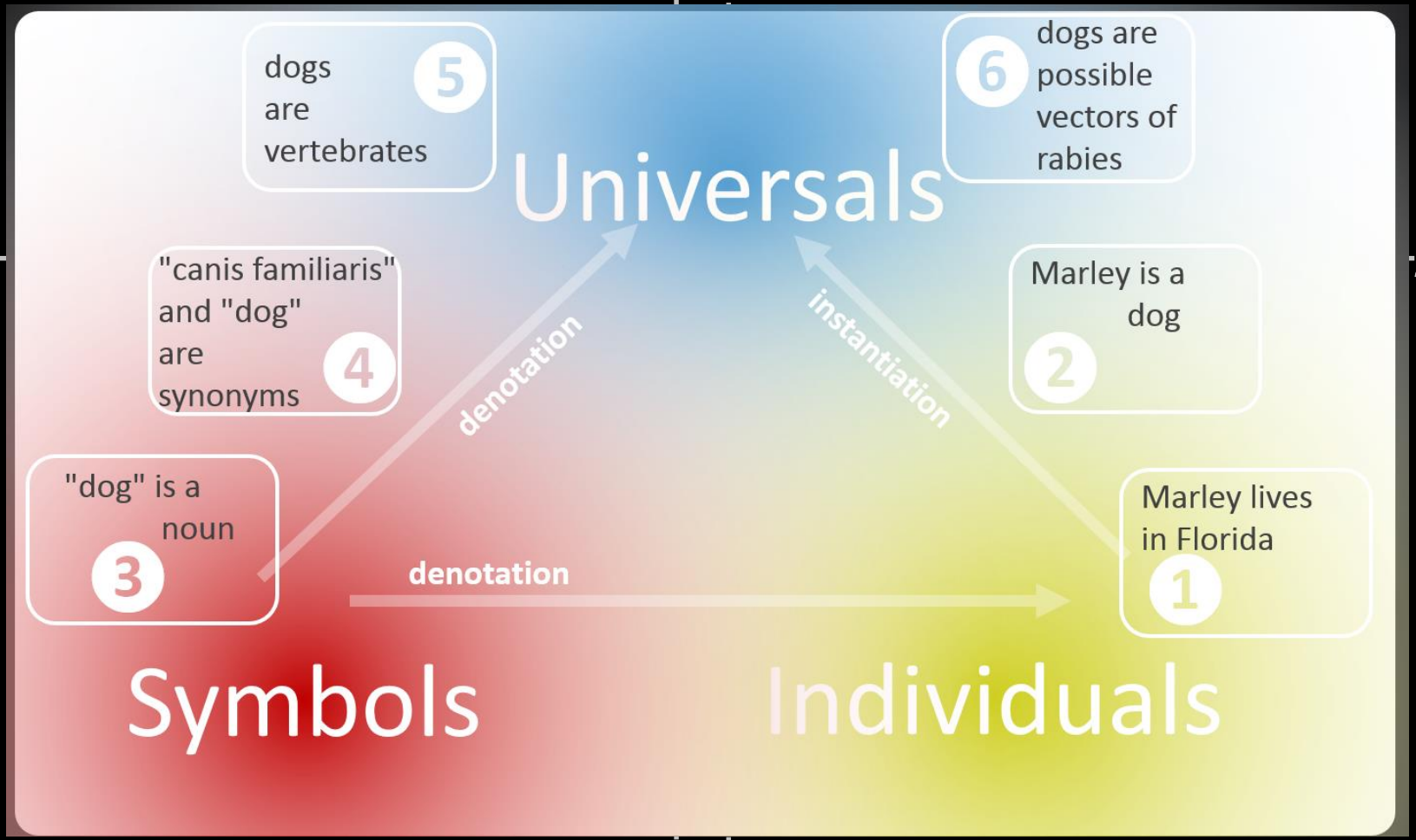

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Ontological knowledge:

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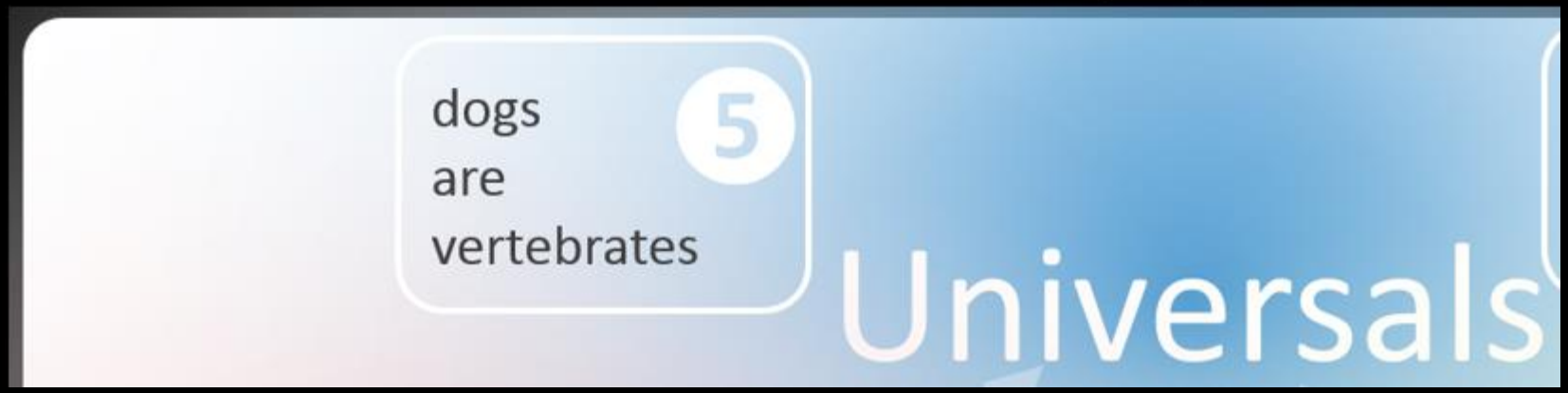
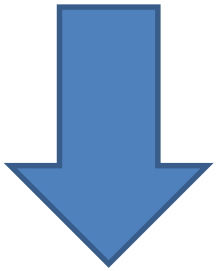


Representation OWL

```
Dog subclassOf Vertebrate
Vertebrate subclassOf Animal
Vertebra subclassOf Bone
Vertebrate equivalentTo Animal and
          has-part some Vertebra
```



computable inference
(e.g. HermiT or Fact++
OWL reasoner)

There is no dog that has no bones

OWL Manchester Syntax: https://www.w3.org/TR/owl2-manchester-syntax/
HermiT reasoner: http://www.hermit-reasoner.com/
Fact++ reasoner: http://owl.man.ac.uk/factplusplus/

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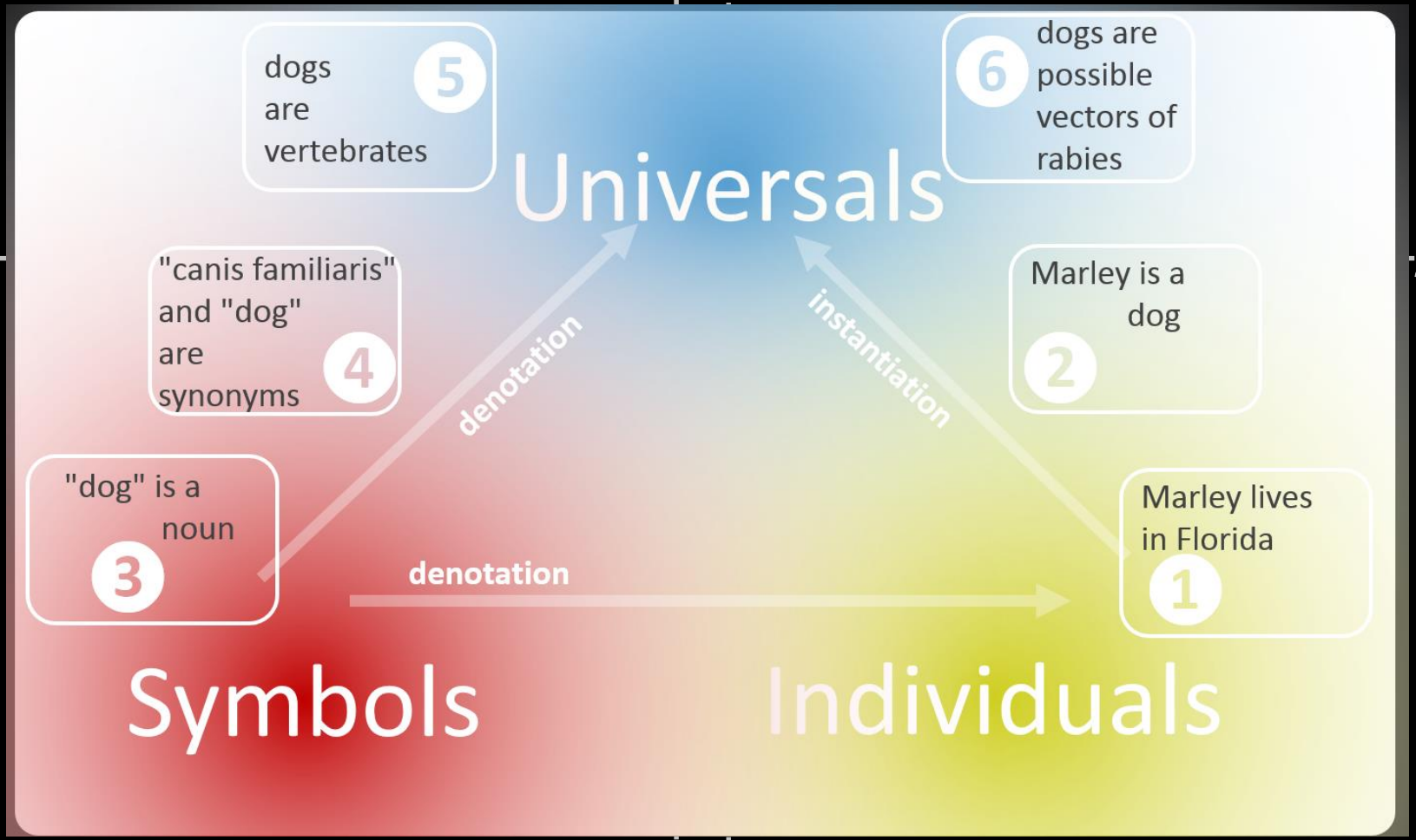
typical, likely, possible



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e

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typical, likely, possible

Triple representation

- No formal semantics!
- Different, mostly complex interpretations
- Don’t use formal languages for this

<Subject>	<Predicate>	<Object>
:Dog	**:vector-of**	**:Rabies**
:Tobacco	**:causes**	**:Cancer**
:Aspirin	**:treats**	**:Pain**
:Fever	**:suggests**	**:Malaria**
:Bird	**:capable-of**	**:Flying**

~~Dog subclassOf vector-of some Rabies~~

~~Tobacco subclassOf causes some Cancer~~

~~Aspirin subclassOf treats some Pain~~

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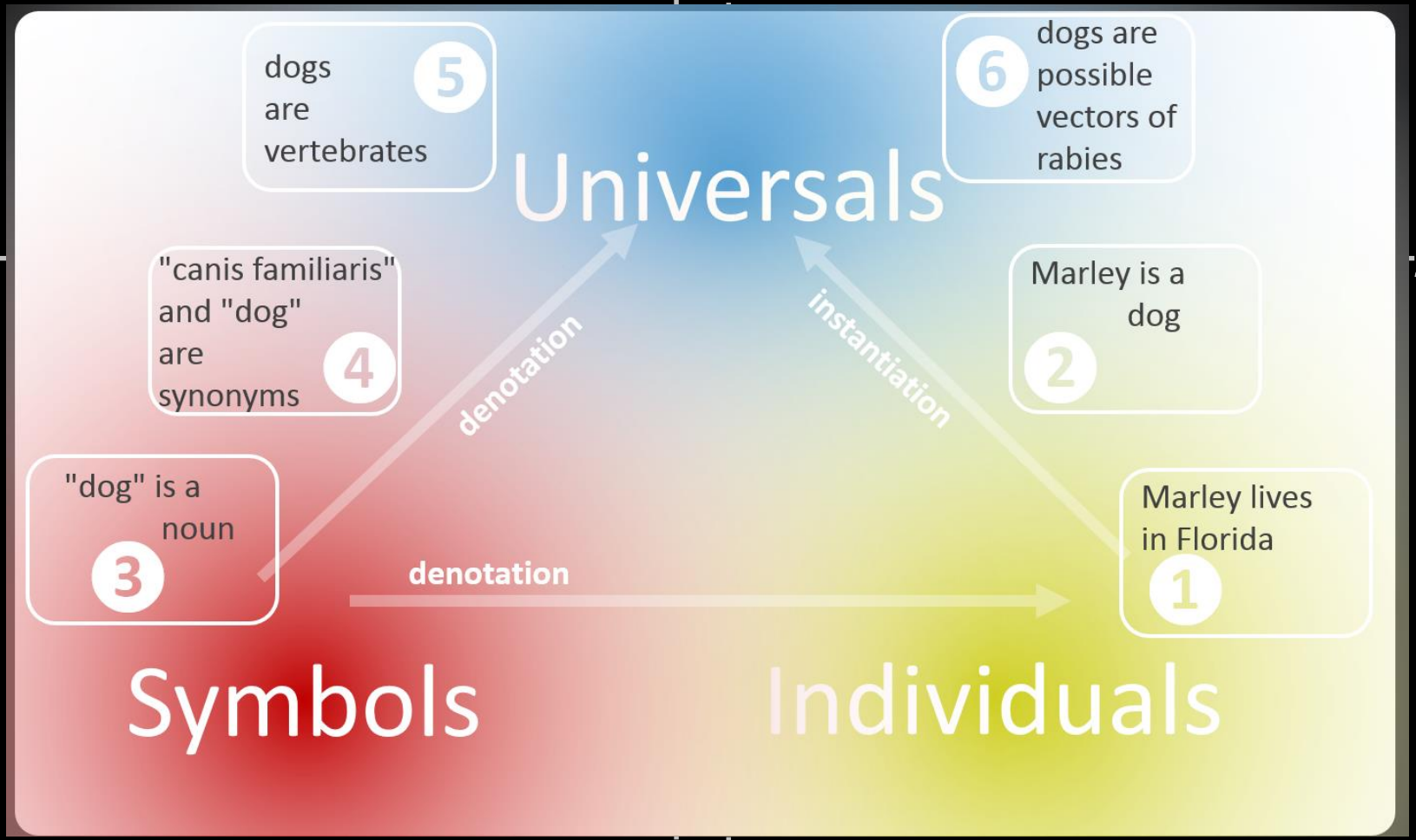

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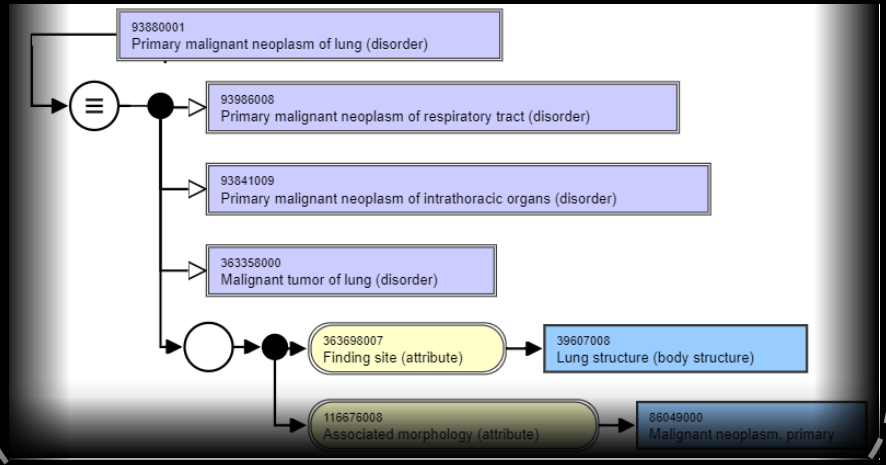
Ontological knowledge

Axiomatic layer of clinical terminology systems



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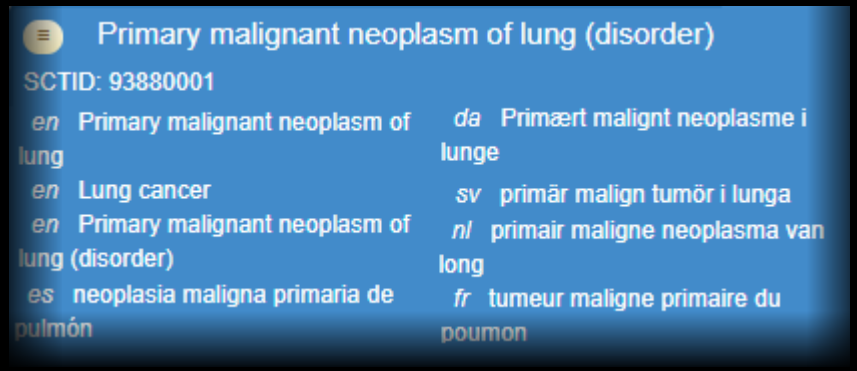
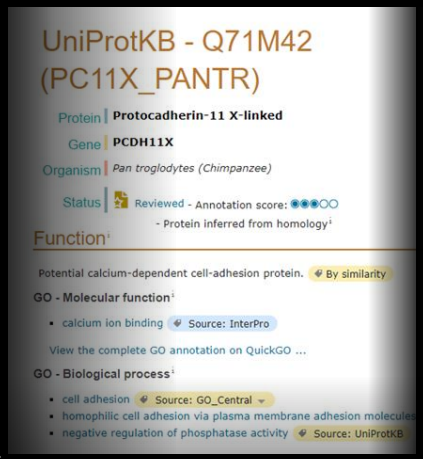


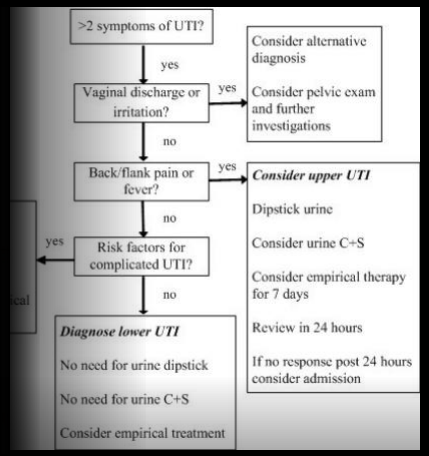
Lexical layer of terminology systems

Symbolic knowledge

Contingent knowledge

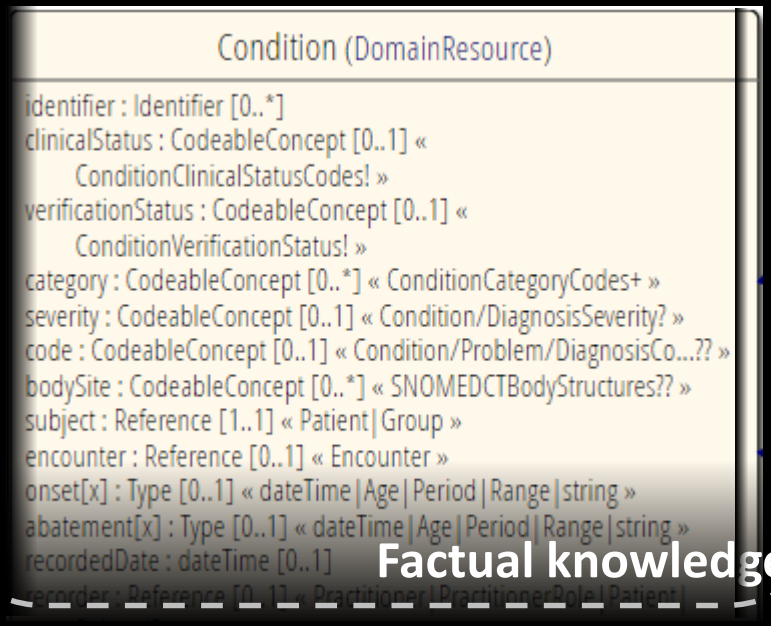
e.g. Research databases Clinical guidelines





HL7 FHIR

e.g. Clinical Information Models



Factual knowledge